Title: Pallister-Hall Syndrome *GeneReview* Table 2

Author: Biesecker LG

Date: September 2012

Note: The following information is provided by the author and has not been reviewed by *GeneReviews* staff.

**Table 2. Selected *GLI3* Putative Normal Allelic Variants**

| DNA Nucleotide Change (Alias) | Protein Amino Acid Change | dbSNP Reference Number |
|---|---|---|
| c.124+61C>A (IVS2+61C>A) | N.A. | |
| c.547A>G | p.His179 | rs3898405 |
| c. 547A>G | p.Thr183Ala | rs846266 |
| c.826+153T>C (IVS6+153T>C) | N.A. | |
| c.900C>T | p.Ser300 | rs35961850 |
| c.963C>T | p.Leu321 | rs34965132 |
| c.1356+11G>C (IVS9+11G>C) | N.A. | rs846273 |
| c.1393G>C | p.Gly465Arg | rs35488756 |
| c.1509C>T | p.Asn503 | rs34020684 |
| c.1728C>T | p.Y576 | rs35128755 |
| c.2361T>A | p.Asn787Lys | rs10259802 |
| c.2408G>A | p.Ala803Val | rs34169786 |
| c.2826G>C | p.Pro942 | rs34245321 |
| c.2835G>C | p.Leu945 | |
| c.2993C>T | p.Pro998Leu | rs929387 |
| c.3774C>G | p.Leu1258 | rs35448119 |
| c.4006G>A | p.Gly1336Glu | rs35280470 |
| c.4020C>T | p.Pro1340 | rs35139358 |
| c.4071C>T | p.Tyr1357 | rs34089404 |
| c.4293G>C | p.Pro1431 | rs28396689 |
| c.4595C>G | p.Ser1532Cys | rs2079451 |
| c.4609C>T | p.Arg1537Cys | rs35364414 |
| c.4705delC | p.Leu1565X | rs35765130 |
| c.*30G>T | N.A. | |
| c.*136T>A | N.A. | |

Most of these variants have been seen in multiple unrelated persons and are not believed to be associated with any phenotypic effects, although they have not been rigorously analyzed for subtle effects. These are included in this table if they lie within an exon or if they are in an intron within 25 bp of an exon.

N.A.= Not applicable.

The dbSNP reference SNP cluster 'rs' ID's are at http://www.ncbi.nlm.nih.gov/SNP/. Readers should refer to dbSNP to confirm these data and for additional data. These SNPs are from the Human Genome build 126. Note: (1) Nomenclature for normal and pathologic allelic variants follows recommendations of den Dunnen & Antonarakis (2001) and the updated recommendations at the Human Gene Variation Society website http://www.hgvs.org/. An asterisk indicates that the variant is located X number of nucleotides after the stop codon; (2) All protein alterations are predicted and not experimentally determined; (3) GLI3 reference sequences are NM_000168.3 and NP_000159.3.